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ABSTRACT

This pamphlet uses a question-and-answer format to provide information about Waardenburg syndrome, an inherited disorder often characterized by varying degrees of hearing loss and changes in skin and hair pigmentation. The pamphlet covers: causes of Waardenburg syndrome. characteristics, types, research being done, ways to help in research projects, and relevant activities of the National Institute on Deafness and Other Communication Disorders. (JDD)

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BECAUSE YOU ASKED ABOUT . . .

WAARDENBURG SYNDROME

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What is Waardenburg syndrome?

Waardenburg syndrome (WS) is an inherited disorder often characterized by varying degrees of hearing loss and changes in skin and hair pigmentation. The syndrome was described in 1951 by Dutch ophthalmologist Petrus Johannes Waardenburg who observed that people with two differently colored eyes often had a hearing impairment. He studied over a thousand individuals in deaf families and found that they had certain physical characteristics in common.

What causes Waardenburg syndrome?

As a genetic disorder, WS is passed down from parent to child much like hair color, blood type, or other physical characteristics. A child receives genetic material from each parent; one gene from the mother and one gene from the father determine the characteristics of a child's particular trait. Because WS is a dominant trait, a child can inherit the syndrome even if only one parent has a WS gene. Therefore, there is a 50/50 chance that a child of an individual with WS will also have the syndrome.

Waardenburg syndrome has many characteristic traits but people with WS may not have all these traits. Sometimes there may only be a few characteristics indicating the presence of a WS gene. Some children of a deaf mother with WS, for example, may have a white forelock and a wide space between the inner corners of their eyes but have no hearing impairment. Others may have white patches of skin and moderate hearing loss.

What are the characteristics of Waardenburg syndrome?

One commonly observed characteristic of Waardenburg syndrome is two differently colored eyes *(heterochromia iridum)*. One eye is usually brown and the other blue. Sometimes, one eye has two different colors *(iris bicolor heterochromia)*. Other individuals with WS may have unusually brilliant blue eyes, often referred to as *Waardenburg blue*.

Patients with WS may also have distinctive hair coloring, such as a patch of white hair or premature gray hair as early as the age of 12. Other possible physical features include a broad nasal root, a wide space between the eyes *(dystopia canthorum)*, confluent or connected eyebrows *(synophrys)*, and a low frontal hair line. The levels of hearing loss associated with the syndrome, which can vary from moderate to profound, are usually non-progressive.

On rare occasions, WS has been associated with other congenital deformities, such as *Hirschsprung's disease*, an intestinal disorder, *Sprengel's deformity*, shoulder blade deformity, and *spina bifida*, which affects the spine. Cleft lip and/or palate have also been associated with WS.

Are there different types of Waardenburg syndrome?

There are two types of WS: Type 1 and Type 2. Individuals who have an unusually wide space between the inner corners of their eyes have WS Type 1. Hearing impairments occur in about 20 percent of individuals with this type of WS. Individuals who have many WS characteristics but do not have a wide space between the inner corners their eyes are described as WS Type 2. About 50 percent of these people are deaf or have a hearing impairment.

What research is being done?

Scientists are trying to find the genes that cause WS using a laboratory technique called linkage analysis. In linkage analysis, scientists examine DNA extracted from blood cells taken from individuals and families affected with WS. DNA, the molecular basis for hereditary traits, is present in every cell in the human body. The DNA for each person is unique, but every cell of a particular person contains an exact copy of that DNA. A gene is a small section of DNA which usually controls one or two traits. Scientists analyze small segments of the DNA to look for the WS genes.

Because family members have similar genes, it is important to include as many family members as possible in the linkage analysis. In general, the more family members tested, the better the linkage analysis.

Genes that are relatively close to one another on the strip of DNA can "link" together and be inherited as a group from a parent. The WS genes may be linked to neighboring genes that express other physical traits. In a step-by-step process, scientists try to locate linkages of WS genes. Current analysis of several families suggests that one of the Waardenburg genes is located on a section of chromosome 2. Chromosome 2 is just one of the 23 pairs of chromosomes which house a person's genes. Analysis of some families, however, suggests that not all WS genes link to this portion of chromosome 2.

Once the locations of the WS genes are found, scientists can develop gene therapy which may prevent or lessen the hearing loss associated with the disease. Scientists believe that further research in genetics will lead to a better understanding of the development of the normal human ear and central nervous system.

What can I do to help myself?

Scientists need individuals and families with WS for linkage analysis studies. If someone in your family has WS, you and your relatives may be able to contribute to the understanding of Waardenburg syndrome. By donating a small sample of your blood, you could help scientists in WS research. For additional information, a patient's physician may wish to contact:

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About the NIDCD

The National Institute on Deafness and Other Communication Disorders (NIDCD) is one of the institutes of the National Institutes of Health. The NIDCD conducts and supports research and research training on normal mechanisms as well as diseases and disorders of hearing, balance, smell, taste, voice, speech and language. The NIDCD achieves its mission through a wide range of research performed in its own laboratories, a program of research grants, individual and institutional research training awards, career development awards, center grants, and contracts to public and private research institutions and organizations.

The Institute also conducts and supports research and research training related to disease prevention and health promotion; addresses special biomedical and behavioral problems associated with people who have communication impairments or disorders; and supports efforts to create devices which substitute for lost and impaired sensory and communication function. The NIDCD is committed to understanding how certain diseases or disorders may affect women, men, or members of underrepresented minority populations differently.

The NIDCD has a national clearinghouse of information and resources. Additional information on hearing impairments may be obtained from the NIDCD Clearinghouse. Write to:

NIDCD Clearinghouse
P.O. Box 37777
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